SLC45A2 gene

solute carrier family 45 member 2

Normal Function

The *SLC45A2* gene (also called *MATP*) provides instructions for making a protein that is located in specialized cells called melanocytes. These cells produce a pigment called melanin, which is the substance that gives skin, hair, and eyes their color. Melanin is also found in the light-sensitive tissue at the back of the eye (the retina), where it plays a role in normal vision.

Although the exact function of the SLC45A2 protein is unknown, it is likely involved in the production of melanin. This protein probably transports molecules necessary for the normal function of melanosomes, which are the structures in melanocytes where melanin is produced. Studies suggest that certain common variations (polymorphisms) in the *SLC45A2* gene may be associated with normal differences in skin, hair, and eye coloring.

Health Conditions Related to Genetic Changes

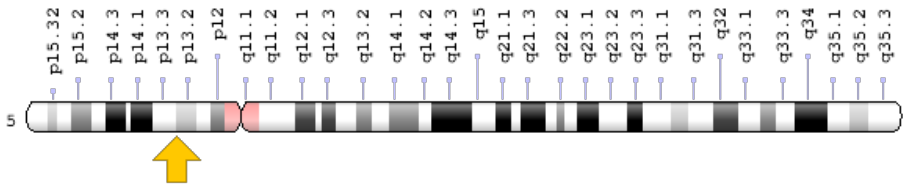
oculocutaneous albinism

More than 20 mutations in the *SLC45A2* gene are responsible for oculocutaneous albinism type 4. The most common *SLC45A2* mutation in the Japanese population switches a single protein building block (amino acid) in the SLC45A2 protein. Specifically, this mutation replaces the amino acid aspartic acid with the amino acid asparagine at protein position 157 (written as Asp157Asn or D157N). Other mutations, including changes in single amino acids and deletions or insertions of genetic material in the *SLC45A2* gene, have also been reported in several populations worldwide. Mutations in this gene reduce or eliminate the function of the SLC45A2 protein in melanin production. Because this protein is important for normal pigmentation, its loss leads to changes in skin, hair, and eye coloration and problems with vision that are characteristic of oculocutaneous albinism type 4.

Chromosomal Location

Cytogenetic Location: 5p13.2, which is the short (p) arm of chromosome 5 at position 13.2

Molecular Location: base pairs 33,944,616 to 33,984,675 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AIM-1
- AIM1
- MATP
- melanoma antigen AIM1
- membrane associated transporter
- membrane-associated transporter protein
- S45A2_HUMAN
- solute carrier family 45, member 2

Additional Information & Resources

GeneReviews

- Oculocutaneous Albinism Type 4
  https://www.ncbi.nlm.nih.gov/books/NBK1510

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MATP%5BTIAB%5D%29+OR+%28membrane+associated+transporter%5BTIAB%5D%29%29+OR+%28%28AIM-1%5BTIAB%5D%29+OR+%28AIM1%5BTIAB%5D%29+OR+%28melanoma+antigen+AIM1%5BTIAB%5D%29+OR+%28membrane-associated+transporter+protein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 45, MEMBER 2
  http://omim.org/entry/606202

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC45A2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC45A2%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16472
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/51151
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